



## AASS gene

aminoadipate-semialdehyde synthase

### Normal Function

The AASS gene provides instructions for making an enzyme called aminoadipic semialdehyde synthase. This enzyme is found in most tissues, with the highest amounts found in the liver. Aminoadipic semialdehyde synthase is involved in the breakdown of the amino acid lysine, a building block of most proteins. It is called a bifunctional enzyme because it performs two functions. One function, called lysine-ketoglutarate reductase, breaks down lysine to a molecule called saccharopine. The other function, called saccharopine dehydrogenase, breaks down saccharopine to a molecule called alpha-aminoadipate semialdehyde.

### Health Conditions Related to Genetic Changes

#### hyperlysinemia

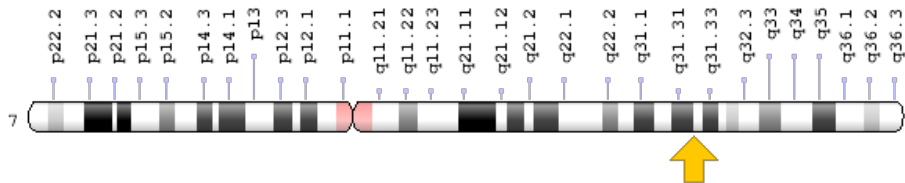
At least five mutations in the AASS gene have been found to cause hyperlysinemia. Most of these mutations change single amino acids in aminoadipic semialdehyde synthase. These mutations are thought to decrease or eliminate enzyme activity, resulting in an inability to break down lysine. Lysine that is not broken down accumulates in the blood, but it typically causes no health problems.

When mutations in the AASS gene impair the breakdown of saccharopine, this molecule builds up in blood and urine. This buildup is sometimes referred to as saccharopinuria, which is considered to be a variant of hyperlysinemia. It is unclear if saccharopinuria causes any symptoms.

## Chromosomal Location

Cytogenetic Location: 7q31.32, which is the long (q) arm of chromosome 7 at position 31.32

Molecular Location: base pairs 122,073,544 to 122,144,290 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- AASS\_HUMAN
- alpha-aminoadipate semialdehyde synthase
- aminoadipic semialdehyde synthase
- LKR/SDH
- LKRSDH
- lysine-2-oxoglutarate reductase
- lysine-ketoglutarate reductase /saccharopine dehydrogenase

## Additional Information & Resources

### Genetic Testing Registry

- GTR: Genetic tests for AASS  
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10157%5Bgeneid%5D>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28AASS%5BTIAB%5D%29+OR+%28aminoadipate-semialdehyde+synthase%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- ALPHA-AMINOADIPIC SEMIALDEHYDE SYNTHASE  
<http://omim.org/entry/605113>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=AASS%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=17366](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17366)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/10157>
- UniProt  
<http://www.uniprot.org/uniprot/Q9UDR5>

## **Sources for This Summary**

- OMIM: ALPHA-AMINOADIPIC SEMIALDEHYDE SYNTHASE  
<http://omim.org/entry/605113>
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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17513445>

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